

# **3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency (HMG)**

An organic acid disorder

## ***What is it?***

3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency (also known as HMG) is an inherited organic acid disorder. People with organic acid disorders, like HMG, cannot properly break down certain components of protein. This is because the body is lacking a specific chemical called an enzyme. Since the body cannot properly break down protein, certain organic acids build up in the blood and urine and cause problems when a person eats normal amounts of protein, or becomes sick.

## ***What are the symptoms?***

A person with HMG can appear normal at birth. The symptoms of HMG can be very variable between people. People may present with vomiting, low blood sugar, low muscle tone, and large livers. Some people with HMG may have no symptoms at all. Many symptoms of HMG can be prevented by immediate treatment and lifelong management. People with HMG typically receive follow-up care by a team of professionals that is experienced in treating people with metabolic disorders.

## ***Inheritance and frequency***

HMG is inherited in an autosomal recessive manner. This means that for a person to be affected with HMG, he or she must have inherited two non-working copies of the gene responsible for causing HMG. Usually, both parents of a person affected with an autosomal recessive disorder are unaffected because they are carriers. This means that they have one working copy of the gene, and one non-working copy of the gene. When both parents are carriers, there is a 1 in 4 (or 25%) chance that both parents will pass on the non working copies of their gene, causing the baby to have HMG. Typically, there is no family history of HMG in an affected person. HMG is a rare organic acid disorder; the number of people with HMG is not known. It is more common in people from Saudi Arabia.

## ***How is it detected?***

HMG can be detected through newborn screening. A recognizable pattern of elevated chemicals alerts the laboratory that a baby may be affected. Confirmation of newborn screening results is required to make a firm diagnosis. This is usually done by a physician that specializes in metabolic conditions, or a primary care physician.

## ***How is it treated?***

HMG is treated by eating a diet low in protein, and sometimes medication, as recommended by a genetic metabolic medical professional.

**DISCLAIMER: This information is not intended to replace the advice of a**

**genetic metabolic medical professional.**

**For more information:**

**Genetics Home Reference**

Website: <http://ghr.nlm.nih.gov/ghr/page/Home>

**Save Babies Through Screening Foundation**

4 Manor View Circle Malvern, PA 19355-1622 Toll Free Phone: 1-888-454-3383

Fax: (610) 993-0545 Email: [email@savebabies.org](mailto:email@savebabies.org)

Website: <http://www.savebabies.org/diseasedescriptions.php/>

**Organic Acidemia Association**

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1797 **Fax:** 763-694-0017 **Email:** [oaanews@aol.com](mailto:oaanews@aol.com)

[www.oaanews.org](http://www.oaanews.org)

**STAR-G Hawaii Department of Health**

<http://www.newbornscreening.info/Parents/organicaciddisorders/HMGCoA.html>